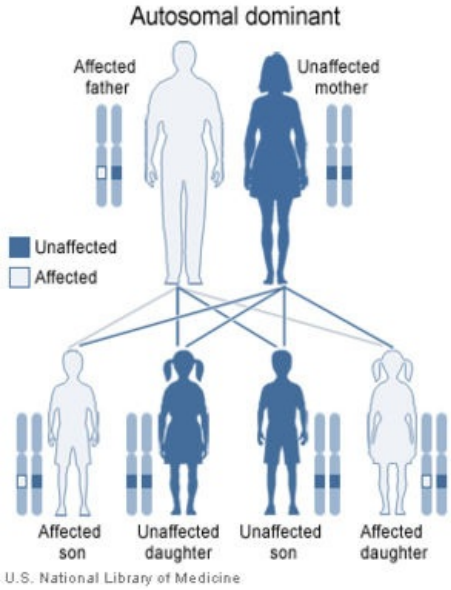
Goldenhar syndrome

Named after the Belgian-born American ophthalmologist *Maurice Goldenhar*.[1]

Genetic essence

- OMIM: 164210 (http://omim.org/entry/164210).
- Inheritance: most cases are sporadic, the described inheritance is autosomal dominant, cases of autosomal recessive inheritance were previously described.[2]



Genetic essence

Characteristic features

- microphthalmia;
- congenital cataract;
- preauricular fistula;
- possibly also atresia of the ear canal;
- unilateral facial hypoplasia;
- macrostomia;
- anomalous position of the teeth;
- overall retardation in psychological development;
- anomalies of ribs and vertebrae.

Links

References

1. ENERSEN, Ole Daniel. *Whonamedit - Maurice Goldenhar* [online]. [cit. 2011-04-23]. <http://www.whonamedit.com/doctor.cfm/2048.html>.
2. *OMIM : Hemifacial microsomia - Clinical synopsis* [database]. Johns Hopkins University. [cit. 2011-04-23]. <http://omim.org/entry/164210>.

Used literature

- LAZOVSKIS, Ilmars – DOBIÁŠ, Václav. *Přehled klinických symptomů a syndromů.* 2. edition. Avicenum, zdravotnické nakladatelství, 1990. ISBN 80-201-0043-1.